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ABSTRACT

This special edition explores the serious genetic disorder, Lesch-Nyhan Disease (LND), which is characterized by severe dystonia, spasticity, speech impairment, renal disease, varying degrees of cognitive deficit, and, especially, compulsive self-injury. The information provided is based on experience at the Matheny School and Hospital (New Jersey) and reflects that institution's philosophy of individualized treatment and development of skills necessary to self-direct one's life, regardless of the degree of disability. In the first section, an overview of LND is provided in tandem with a column of observations and comments by staff at the school about a specific case. Later sections focus on self-injurious behavior, health care, adapted equipment, speech/communication/feeding, education, and leisure/recreation. (Contains 11 references.) (DB)

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SPECIAL EDITION
LESCH-NYHAN DISEASE

From time to time the Matheny Bulletin will depart from its general literature review format in order to present a discussion on a particular topic. This Special Edition explores a rare and devastating genetic disorder, Lesch-Nyhan Disease (LND). LND is characterized by severe dystonia, spasticity, speech impairment, renal disease, varying degrees of cognitive deficit, and the hallmark symptom, compulsive self-injury. The Matheny School and Hospital has been providing residential programs for individuals with this condition for two decades and currently seven boys and young men with LND reside at Matheny. Considering the low incidence of LND, this represents a relatively large population for a single residential facility. Matheny has also provided out patient services and respite care services, and runs an ongoing LND parent support group.

Because of this long experience with LND, Matheny staff are frequently called upon to provide consultation to service providers who have encountered their first case, and to parents of boys with LND. On such occasions we have found that it would be helpful to have written materials, but very little has been published on this condition and most of what has been published is highly technical and focuses on genetic and biochemical issues. It is the infrequent article that deals with the clinical aspects of the disease, whether medical or behavioral, and these papers also tend to be research oriented and of little practical help to those dealing directly with a student, patient, or child with LND.

The intent of this Special Edition of the Matheny Bulletin is to create an informative document that will serve to introduce the uninitiated reader to LND and its unique symptoms, and to offer practical suggestions for those providing direct services. The information is drawn mostly from Matheny's direct care experience and reflects a philosophy that stresses individualized treatment and the development of skills necessary to self-direct one's life, regardless of the degree of disability.

OVERVIEW OF LESCH-NYHAN DISEASE

Lesch Nyhan-Disease (LND) was first reported by Lesch and Nyhan in 1964[1] when they described two affected brothers. LND is a rare condition that is caused by a defective gene on the X chromosome. The condition can be inherited, or can occur spontaneously via a genetic mutation[2]. Since the defective gene is recessive, females almost never exhibit the disease, but may be carriers. There are only two documented cases of females with LND in the world's literature and various mutations have been hypothesized for this rare occurrence[3]. LND appears to be distributed evenly among races and geographic locales and occurs in approximately one of every 380,000 births[4]. As a consequence there are only several hundred individuals with LND currently living in the United States.

LND is associated with a nearly complete absence of the enzyme hypoxanthine guanine phosphoribosyl transferase (HGPRTase), which metabolizes hypoxanthine and guanine to uric acid.

The block in this metabolic pathway leads to an accumulation of uric acid in the blood (hyperuricemia) with the precipitation of uric acid in the urine. Untreated, it generally leads to gradual and fatal injury to the kidneys. The most significant advance in altering the natural history of LND has been the use of allopurinol to decrease serum uric acid production and rate of nephropathy (renal injury). The medication has been utilized for approximately two decades so it is

This column contains a variety of comments and observations made by people who work closely with one or more boys with Lesch-Nyhan disease. A fictitious boy, John, is utilized to represent the actual boys being referenced.

Observation (7:00 AM): John is being transferred from bed to wheelchair. Two personal care assistants (PCA's) are needed. One will lift and control John, the other will secure his seat belt and various straps. A hand hits one of the PCA's in the face. John says, "Sorry, sorry, sorry." The PCAs continue as if nothing has happened.

Parent: "I know I shouldn't say this, but sometimes I wish I had a child with just a regular disability like cerebral palsy. This is so hard."

Observation (7:30 AM): Breakfast time for John begins with a choice of food. His PCA asks, "What would you like, eggs or cereal?" John chooses cereal. The PCA repeats, "Cereal?" John replies, "No." During breakfast John eats with two other students. The PCA asks, "What are you all doing today?" John answers "School." The PCA says, "What will you do in school?" John spits his cereal at the PCA.

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still too early to tell to what degree it can prevent significant renal injury over time and prolong life in affected individuals. However, children are no longer dying in their early teens from uric acid nephropathy. It is important to keep in mind that allopurinol does not impact on the devastating neurologic manifestations of the disorder which are the result of a profound derangement in the metabolism of purines required for normal brain functioning. There is evidence that the abnormality in purine metabolism leads to an imbalance in neurotransmitter (chemicals needed for brain cells to function properly) metabolism in the central nervous system. It is felt that this derangement in neurotransmitter metabolism leads to the behavioral and neurologic manifestations of the disorder. Most studies have focused on abnormalities in dopamine function and cerebrospinal fluid levels of dopamine metabolites appear to be decreased in individuals with LND[6].

It is remarkable to find that when computerized axial tomography (CAT scan), or magnetic resonance imaging (MRI) of the brain is performed in LND patients, the studies are normal. If structural abnormalities exist they are most likely at a microscopic level and, as a consequence, studies that assess the gross appearance of the brain tend to be normal. There have been very few post-mortem examinations of the brains of individuals with LND. A detailed histopathological and electron microscopic examination of the brain in one case revealed no abnormalities[6]. Studies utilizing positron emission tomography are currently underway to identify whether regional metabolic abnormalities can be identified.

Clinical Course: At birth, children with LND appear normal and gross motor milestones may be achieved appropriately up to six to eight months of age in occasional individuals. As a consequence, some children may learn to sit and transfer from one hand to the other. Between 8 and 24 months of age, however, choreoathetosis (an involuntary twisting movement) develops and there is a loss of early milestones. At first, infants are hypotonic (have decreased muscle tone), but later they develop hypertonia (increased tone) and hyper-reflexia.

By four years of age many of the affected children begin to exhibit the classic manifestation of LND, self-mutilation. The self-mutilation seen in LND is generally quite severe and can lead to the loss of lips and fingers from biting, visual loss from rubbing the eyes, and any number of other injuries. It is hypothesized that the self-mutilation is related to neurotransmitter abnormalities, in particular derangements in serotonin or dopamine metabolism. It is of interest that self mutilation can also be seen in other conditions such as autism, Cornelia DeLange Syndrome, Tourette Syndrome, and Down Syndrome. In these conditions, abnormalities in serotonin metabolism have also been hypothesized.

Observation (8:30 AM): John goes back to his bedroom to get ready for school. His PCA says, "I need to wash your hands and face." John replies, "TV." The PCA asks, "What channel?" John answers, "Eleven." John's face is washed without incident while he is engrossed in a program.

Physician: "When I first examined John, I started to take his arm restraint off. He immediately yelled, "It's not safe!" and did not relax until a therapist who was well-known to him came in to assist with the exam."

Observation (9:15 AM): While being wheeled to school John extends his arms outward when going through a doorway and yells "Ouch!"

Parent: "One time my nose was broken when we were trying to get my son dressed. He is quite strong when he throws his head back in extension."

Observation (10:00 AM): John is filling out a permission slip for a trip to the bank. He uses a single switch to scan through choices presented on a computer screen. John says, "Help me." The teacher asks John, "Where are you going?" He replies, "Bank." The teacher explains that he should hit the switch when it scans to the word BANK. John hits the switch when it is on GROCERY STORE but says, "I don't mean that." The teacher deletes the response and John tries again. This time he hits the correct word. The teacher says, "Great, what's next?" John says, "No more."

Observation (11:00 AM): John's occupational therapist and rehabilitation engineer need to measure him for a piece of equipment. John is very upset, crying and screaming.

Observation (12 Noon): John is transported to the dining room for lunch. The teacher bends over to put John's brake on and John grabs her hair and pulls. The teacher's head is smashed into the wheel of the chair. She tries to release John's grip but he holds on tightly.

The *Matheny Bulletin* is a selective review of papers from the world literature on cerebral palsy and other chronic neurologic conditions. It is an educational service provided to physicians, nurses, therapists, educators, and other professionals involved in the care, management, and education of children and young adults with handicaps. The *Bulletin* is designed to stimulate thought and to further investigation and does not provide advice regarding the diagnosis or treatment for any individual case. Please direct all questions and comments to: Gabor Barabas, M.D., Editor, *Matheny Bulletin*, Matheny School and Hospital, Peapack, New Jersey 07977, (908) 234-0011. Matheny is a school and hospital that serves the needs of children and young adults with severe physical disabilities.
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By eight to ten years of age almost all children exhibit self-injurious behavior and demonstrate the neurologic manifestations of the disorder including spasticity, choreoathetosis, opisthotonos (arching of the back), and facial dystonia. The increase in muscle tone frequently leads to scissoring (crossing of the legs) and children are unable to sit or ambulate. The neurologic manifestations of LND impact profoundly on independence and, to varying degrees, individuals are dependant on others for self-care. The dystonia of facial muscles as well as what appears to be a bucco-lingual dyspraxia (incoordination of lips and tongue) lead to poor feeding and problems with coordination of swallowing. Many individuals have recurrent vomiting, and chronic "silent" aspiration can result which may lead to gradual pulmonary and nutritional compromise. In almost all cases physical growth and development are significantly retarded, for it is extremely difficult to feed children and adults with athetosis and dystonia who have vomiting and dysphagia (difficulty swallowing). As communication develops, a severe disturbance in speech (dysarthria) is observed in association with a delay in language. Most affected children, however, appear to comprehend quite well.

Formal IQ testing in LND generally reveals deficits in intellectual capabilities. Most individuals test within the mild to moderate mentally retarded range. However, those who work with and interact with these children and adults on a day to day basis will generally comment that these results tend to under-estimate their "true" intellectual capabilities. It has been suggested in some reports that there is a gradual deterioration in IQ over time, but there have been no formal or well constructed studies to demonstrate that this is the case. IQ's generally range from 50-85 although there have been individuals described with higher IQ's. Because of the serious behavioral and motor impairments in LND, refined psychological testing is generally limited.

Adult males with LND appear to have impaired reproductive capacity and secondary sexual development, and it is likely that there are neuroendocrine abnormalities, as yet unidentified, that are operative.

Diagnosis: The presence of high uric acid in the urine frequently leads to the excretion of orange crystals in diapers. Many children have been first suspected to have LND when orange discoloration of their diapers is noted by their families. The presence of an elevated serum uric acid level and "orange sand" in a child's diaper make the diagnosis of LND quite probable. Definitive diagnosis is made by assaying HGPRTase levels in red blood cells or cultured fibroblasts. It is also possible to identify in utero cases by culturing amniotic fluid fibroblasts and assaying for the deficient enzyme[7].

Therapy: The prevention of renal injury has already been described. Various medications have been utilized to decrease the abnormalities in tone and the involuntary movements. Thioridazine (Mellaril) and haloperidol (Haldol), as well as other drugs, have been tried. The experience with these drugs has been less than satisfactory and no significant impact on the debilitating symptoms has been observed. In essence, there is no known pharmacologic or therapeutic modality at this time that significantly alters the neurologic impact of LND. Various medications have been prescribed to modify the self-mutilation. There have been reports that administering 5-hydroxytryptamine and carbidopa can decrease symptoms. In one report there was transient improvement but the study has not been reproduced successfully[8]. It is quite likely that LND will be one of the target disorders that may benefit from gene therapy in the future. The involved gene has been cloned and sequenced[9] and the polymerase chain reaction technique has permitted the precise identification of alterations in the sequences of bases that result from point mutation. Bone marrow transplantation has been attempted but has been unsuccessful[10].

Observation (12:30 PM): Lunch begins with John asking for a drink. John continues to perseverate on drinking. Before he has finished half his food he says, "No more."

Personal Care Assistant: "I asked John if he wanted to watch his favorite show and he yelled 'No!' I know he wanted to watch it but I think he was punishing himself."

Observation (1:15 PM): John is in his bedroom doing his activities of daily living (ADL's). When being transferred into his wheelchair, John begins screaming, extending, and snapping his head forward.

Observation (1:30 PM): John is back at school in Current Events class. The teacher asks if anybody would like to read a part of a newspaper. John replies, "TV Guide!" The teacher asks, "What show?" John answers, "Bill Cosby." The teacher asks John to read the description of the show. John's volume increases as he reads the show's time and channel. John then says, "BM, BM." He next begins to read the description but stops and says, "No more."

Observation (2:00 PM): John is on a trip to the grocery store. He has his own money and would like to buy a soda and cookies. The speech therapist pushes John's wheelchair while he directs where he would like to go. John gets excited in the candy aisle naming various candy bars. He is able to locate the soda and cookies and make his choices. When checking out John reaches his hand into a display rack and knocks over the items.

Personal Care Assistant: "When John sees me he asks, with apparent hope, if I will be his PCA that night. Then when I answer 'Yes' he curses at me."

Observation (4:00 PM): John participates in International Cooking group. While mixing the batter he knocks the bowl off the table.

SELF-INJURIOUS BEHAVIOR

It is the compulsion to self-injure that represents the most difficult management issue of LND. This behavior usually starts early in life, generally by 4 years of age, but may manifest at a later age. The behaviors seem to escalate as the child grows and becomes more physically capable of inflicting self-injury, and as he becomes more cognitively capable of conceiving new methods of self-injury. The first manifestations of self-injury are usually lip biting, finger biting, and biting of the buccal mucosa (inside of the cheek). This biting can be of such severity that partial amputation of fingers and lips may occur, and can become life threatening because of superimposed infections and tissue necrosis. Extraction of teeth is frequently necessary. Other early forms of self-injury are head banging, arm and leg banging, rubbing various body parts till raw, nose gouging, and eye gouging. In most cases some form of mechanical restraint becomes necessary to protect the individual from himself. Indeed, designing comfortable and functional restraints that do not become further instruments for self-injury is an ongoing challenge. For example, it has been documented that self-injurious behavior may continue while the individual is sleeping. For this reason night positioning devices, such as body vests that are tied to bed rails, are employed to keep the individual centered in bed so he cannot bang his head, arms, or legs on the rails, or hurl himself out of bed. However, any design must also consider the possibility that the vest itself, with its cords and fasteners, may be used to rub against or even to attempt self-strangulation. Later in life, more subtle forms of self-injury occur, probably motivated by the relative success of the protective restraints. For example, a boy who is wearing an elbow splint to keep his arm straight to prevent biting of the fingers may extend his arm as he is being wheeled through a doorway so as to dislocate a shoulder or break an arm.

In addition to the myriad forms of physical self injury seen in LND, there is another realm of behavioral abnormality. This is discussed as aggressive behavior in much of the Lesch-Nyhan literature, but will be referred to here as indirect self-injury or emotional self-injury. Individuals with LND not only demonstrate a compulsion to injure themselves, they also compulsively attempt to injure or otherwise abuse others, including those they care for the most, such as parents, teachers, and aides. Examples are kicking and head butting while being dressed or bathed, cursing without provocation, spitting or vomiting on care providers, and spilling drinks. After performing these behaviors, the individual will characteristically apologize profusely, only to shortly thereafter repeat the behavior. At Matheny, we have come to recognize these behaviors as indirect attempts at self-injury. These aggressive behaviors cause others to become angry, which may lead to the perpetrator being punished, or, minimally, feeling guilty. In any case, this behavior can be interpreted as a form of self-injury, as the intent may not be to harm the other person, but to suffer the consequences. Such emotional self-injury can be exceedingly subtle. For example, one boy was known to enjoy going to the movies but could not resist the compulsion to answer negatively to an invitation, thus denying himself a favored activity.

It seems reasonable to conclude that individuals with LND do not respond to pain and/or punishment the way others do. Indeed, experiments have demonstrated that these individuals will increase the frequency of a behavior for which they receive a painful electric shock despite the fact that their ability to sense pain appears to be intact[11]. They scream and cry out just as loudly as anyone else when hurt, yet they are unable to stop themselves from behaving in ways that result in pain, both physical and emotional.

Punishment in any form will almost always result in an increase in the undesirable behaviors. Any negative feedback will make matters worse. Simply saying "Stop" will escalate a behavior. Even neutral attention to the behaviors will tend to increase their frequency. For example, saying "Oh, did you hurt yourself?" when a boy bangs his arm will focus attention on that behavior and increase its frequency.

The most appropriate general purpose approach to dealing with Lesch-Nyhan self-injurious behaviors is to employ protective devices. The use of protective devices with LND is much different than the use of physical or chemical restraints in other instances of self-injurious behaviors. Individuals with LND desire the use of protective devices. They do not want to hurt themselves or others, but they know that they will if they are allowed to do so. They become extremely upset and fearful when left unrestrained or unprotected. While restraints are usually viewed as restrictive, in the case of LND, well engineered protective devices are enabling. They reduce the stress and fear of self-injury and allow the individual to concentrate on constructive activity. Most individuals with LND can, to varying degrees, learn to direct the application of their protective devices. Those individuals who do not need the devices at all times can be taught to request them when they are feeling stress and are thus more likely to attempt self-injury. They can also learn to monitor the condition and adjustment of the protective devices. For example, if an arm splint is frayed, or placed on the wrong arm, the individual will communicate that there is a problem. Depending on cognitive and communicative capabilities, this may be indicated by direct reference to the problem, or by a diffuse reaction such as fussing or crying. Care providers should always listen to

these concerns and immediately correct the problem. If the individual with LND loses trust in the care provider's ability to keep him safe, the increased stress will result in a higher frequency and intensity of self-injury.

While the use of protective devices is necessary much of the time, it is also desirable to encourage at least some activity where protection is not dependent upon mechanical devices. There are two major reasons for this. First, there are certain activities such as dressing, bathing, transfers, etc. that require the removal of protective devices. If an individual with LND becomes too dependent on these devices, such activities may become more stressful. The individual needs to know that when an experienced person is attending one to one, he will not be allowed to hurt himself. Second, although the compulsion to self-injure will probably always remain, it is possible to learn simple strategies of self-control that will allow for participation in some activities with minimal protection for short periods of time. The most effective intervention for developing these strategies is positive reinforcement of mutually incompatible behaviors. Affected individuals respond to positive feedback and other rewards for doing constructive activities. The trick is to increase behaviors that do not allow for the simultaneous performance of self-injurious behaviors. For example, if playing music on an electronic synthesizer, sorting clothes, or simply folding hands on his lap are rewarded, these behaviors increase while other behaviors that require hand use, such as finger biting and arm swinging decrease, all without directly addressing the "Lesch-Nyhan behaviors."

There is much discussion in the Lesch-Nyhan literature regarding the use of extinction as a behavior modification intervention to reduce self-injurious behaviors. Extinction calls for the removal of all reinforcement, especially attention, while the maladaptive behavior is being performed. To be effective, the subject must be allowed to perform the behavior repeatedly without any reinforcement or other intervention until the frequency approaches zero. For example, to extinguish finger biting the subject would be allowed to bite the fingers without restraint or any other intervention until the behavior ceased. The risks involved in implementing an extinction procedure for Lesch-Nyhan self-injurious behaviors should be obvious. Severe and permanent physical injury could result. Furthermore, the rate of initial success, long term success, and generalization (reductions in self-injurious behaviors not specifically treated by extinction procedures) are not well established. For these reasons, classic extinction procedures are not recommended as a general approach to the management of Lesch-Nyhan self-injurious behaviors.

However, a less formal and less risky variation of the extinction procedure is appropriate in all settings and is highly recommended as the general approach to interacting with individuals with LND. For our purposes this will be called selective ignoring. Assuming that the appropriate protective devices are in place, or that a hand over hand activity is being conducted, the goal is to act as if the "Lesch-Nyhan behaviors" are not happening, and to continue to conduct business as usual. At no time should there be any verbal recognition of any "Lesch-Nyhan behaviors." For example, if in the middle of an individual reading lesson the boy curses or spits at the teacher, the teacher should continue the lesson without any response to the behaviors. There should be no facial expressions of disapproval, no physical retreat, nor any reassurance such as "That's O.K.", and there certainly should be no punishment. Any response will tend to increase the unwanted behaviors, but if the behaviors are completely ignored, they may decrease. When there is no negative consequence to behavior such as cursing or spitting, the behavior tends to lose its self-injurious value. Even if the behavior persists, at least the educational process continues and desirable behaviors may be recognized and rewarded. So, even though the boy spits or curses during the reading lesson, he also may be reading some words correctly. The attempt at reading should be praised and encouragement given to continue.

There may be times when it is impossible to completely ignore a "Lesch-Nyhan behavior." This is generally when physical injury is imminent or in progress, either to one's self or to another. For example, if the hands are unrestrained during a finger painting activity and the boy attempts to bite his fingers, this cannot be ignored. However, only minimal necessary attention should be given. There need be no verbal response. The teacher should not say "Stop" or any other command. The approach is to intervene as quickly and as unemotionally as possible by grasping the wrist and guiding the hand back to the work surface for continued finger painting. Any verbal interaction should revolve around the painting. The teacher may say, "This is starting to look really nice."

The technique of selective ignoring requires practice and self-discipline. It is not easy to continually monitor and control one's normal responses to abnormal behavior. This is the challenge of working with individuals with LND. It is the key to providing successful programs and assistance. In the simplest terms, the idea is to ignore the "Lesch-Nyhan behaviors," but not the individual. If this is done consistently, people will find that beneath the array of strange and paradoxical behaviors resides an individual who would like nothing better than to fit comfortably into his home or school setting. The staff at Matheny have found these students to be, in their unique ways, friendly and caring individuals. They have the same aspirations and enjoy the same opportunities as other students and are well liked and appreciated for their dynamic personalities.

HEALTH CARE

The primary goal of health care for individuals with LND is to maintain safety and prevent renal disease. Families and health care providers have a tendency to manage the medical aspects of LND on a situation-by-situation basis. However, a preventive model is potentially more beneficial.

For young boys with LND, outstanding health issues may interfere with recommended immunization practices. If immunizations are incomplete the situation should be corrected in as timely a manner as possible. In general, individuals with LND are not more susceptible to infections than others, but since abrasion injuries and ulcerations are common consequences of self-injury, secondary wound infection may result. Care for abrasion injuries must be prompt to avoid ulcerations due to compulsive rubbing. Not only should abrasion injuries be treated promptly, but an investigation regarding the environmental cause of the abrasion should be conducted. Changes or additions to the individual's protective equipment need to be made as quickly as possible to avoid further injury. While family members and personal care providers often become excellent designers and fabricators of these special devices, it may be necessary to consult with an equipment specialist, such as a rehabilitation engineer, occupational therapist, or physical therapist. Local rehabilitation hospitals are often valuable resources for such services. Creative thinking also helps. A wrestling helmet was found to be useful for one boy who had chronic rubbing of the ear against various surfaces of his wheelchair seating system.

Minimizing the formation of kidney stones is a significant medical challenge. Allopurinol therapy can substantially inhibit the development of uric acid stones, but in our experience, xanthine stones may form if uric acid levels are too low. To prevent stones, a daily intake of up to 40 ounces of fluids should be provided. Questions should be asked about an individual's fluid preference and pattern of intake (type, time, amount). When encouraging fluids, excessive milk should not be offered, for one boy developed a calcium stone when milk was his primary fluid. Kidney stones may be more prevalent in the summer or early fall when dehydration is more likely. An air-conditioned environment is helpful and extra fluids should be offered when temperatures are high. Alkalinizing the urine with polycitra or bicarbonate keeps the urine neutral and minimizes stone formation. Due to the risk of renal stones, it is recommended that persons with LND see a kidney specialist periodically. A suggested "kidney protocol" is outlined below:

DAILY:	Encourage about 40 ounces of fluids per day, which may necessitate offering of fluids in the classroom. Because of vomiting, frequent small amounts work best.
3/WEEK:	Obtain urinalysis with specific gravity and pH assessment. If the specific gravity is greater than 1.020 or the pH is less than 6.5 or greater than 7.5 over several readings, increase fluid intake and correct acid/base status.
4/YEAR:	Monitor BUN, Serum Creatinine, Electrolytes.
YEARLY:	Renal ultrasound.
YEARLY:	Creatinine clearance; citrate, uric acid and calcium/creatinine ratios.

Kidney ultrasound appears to be the superior imaging study for monitoring kidney growth, progression of stone size, and development of hydronephrosis. Virtually all patients with LND after the age of three have tiny stone deposits in the kidneys at all times, despite appropriate allopurinal therapy. Stones rarely become symptomatic unless fluid status or urine pH are consistently abnormal. Gout is rarely seen.

Bladder and bowel habits need to be carefully assessed. While toilet training may be a possibility for some individuals with LND, many have great difficulty with this expectation. The goal should be to help the individual keep dry, clean, comfortable, and safe, regardless of the method. If a standard toilet fixture is used, transfers must be done with care to avoid self-injury or accidental injury in the inherently dangerous bathroom environment. Special seating equipment may be necessary for positioning and/or restraint. Alternative methods are the use of a hand held urinal, bed pan, diapers, or a combination of the various options. Compulsive "Lesch-Nyhan behaviors" may appear during bladder and bowel routines, whatever the method. One young man was noted to ask for a urinal and when provided with it, would not use it. He would then urinate when it was taken away. In such circumstances it may be more adaptive to use diapers while the compulsive behavior persists. As in all activities, the best approach is usually the one that minimizes stress.

Many boys with LND have recurrent vomiting. Although this is thought to be a symptom of the compulsive, self-injurious behavior, esophageal dysmotility (as measured on manometry) has been found in two children studied at Matheny. Due to the slow movement of food through the upper gastrointestinal tract, these boys can readily bring up food and precipitate vomiting.

The need for tooth extraction to reduce self-injury is common. Primary teeth frequently need to be removed, but some secondary molars can be saved occasionally since oral self-injury tends to be less common in older children. For those patients who have had their teeth removed, a chopped diet is necessary. It should be noted that mouth guards have been found to be of only limited benefit in preventing self-injury and may cause further damage.

ADAPTED EQUIPMENT

Virtually all boys with LND require wheelchairs for mobility. In order to ensure correct positioning, wheelchair inserts are customized to provide optimal support and reduce deforming forces. For some individuals, devices such as wrist cuffs are utilized to enable safe use of a manual language board or other lap board equipment. The cuffs are either attached directly to the lap board or to straps tied to the frame of the wheelchair. When correctly designed, a wrist cuff system allows functional use of the hands in the area of the lap board, while limiting the range of movement to prevent use of the hands and arms for self-injury. Soft elbow splints fastened with Velcro may sometimes offer an alternative solution. By preventing bending of the arm, self-injury in the area of the face is avoided, and the padding protects the arm from banging injuries. Similar systems are frequently needed for the legs as well. Padded foot boxes may be utilized to replace standard wheelchair foot plates.

Besides the common forms of restraints and protective devices employed to provide functional positioning and minimize self-injury, it is often necessary to create unique devices for specific individuals. For example, one boy who enjoys playing Nintendo has a custom lap board designed just for that activity. It has wrist cuffs attached to rings that slide across a raised pipe, giving support to the arms and hands while restricting movement to the area above and around the joy stick.

In addition to the wheelchair, environmental modifications are generally necessary in the bedroom, bathroom and other settings where the individual spends time. Bed vests are made to maintain positioning in bed and to prevent the hands from reaching the mouth. Padded bed rails also help prevent self-injury at night. Customized bath chairs are built to help care providers maintain control of the individual during baths, for bathing is particularly stressful for individuals with LND, probably because the environment is inherently dangerous and leaves no room for care provider error. At our facility, it is common practice to assign two personal care assistants to bathe each individual with LND.

Adapted equipment for individuals with LND must be strong and at the same time smooth and soft, and free of sharp edges or heavy seams in the material. A nylon webbing is used for much of the equipment (arm splints, seat belts, foot straps, wrist cuffs, chest vests). The webbing is very strong but also rough. When used, it must be hidden or sandwiched between layers of foam. A material that is increasingly used is Rubatex, a rubber-based material which is strong enough and soft enough to replace the nylon webbing in some cases. Rubatex also stretches, which is of further benefit to the individual. Equipment that is strong, flexible, and snug fitting provides the greatest degree of comfort and security. It leads to a greater feeling of safety and a decrease in self-injurious behaviors. An example is a Rubatex bed jacket. One boy previously had a denim jacket to keep him secure, but it allowed him to rub his chin and nose on the rough seams. A Rubatex jacket, which has no seams or sleeves, replaced this and has provided comfort and safety in bed. Although Rubatex is not as strong as denim it is strong enough to keep a boy in place. The one drawback of Rubatex is that it can be warm and lead to increased sweating.

Transportation of individuals with LND is best done in their wheelchairs and with a van equipped with a wheelchair lift. Younger children may be transported safely in cars when their wheelchair insert can be easily removed and used as a car seat. Basic principals regarding safety in transporting individuals in wheelchairs must be strictly followed.

When deciding on what equipment to use, or when measuring individuals with LND for equipment, their unique behavior style must be taken into consideration. It is important to be positive and confident about a change in equipment or severe anxiety can result. One boy injured his hip and required hospitalization due to a sudden severe extension when being fitted for new equipment. Only when the individual has become comfortable working with the treating team, can he fully participate in equipment trials. The best outcome is found when the team is consistent and encouraging. If the individual senses that the team is unsure as to the equipment, he will also be unsure and will tend to sabotage the equipment trial. The team (therapists, teacher, rehabilitation engineer, family, care providers, etc.) must keep in close communication regarding the

needs of particular individuals and the consequent design, fabrication, and utilization of adapted equipment. Intervention must be adjusted as needs change. Through the incorporation of efforts from different parties the individuals will have enhanced functioning as well as ensured safety.

SPEECH/COMMUNICATION/FEEDING

Students with LND present with varying degrees of communication difficulties. In general, receptive language skills appear to be within functional limits. Expressive language is always affected and moderate to severe dysarthria (speech impairment) is exhibited. The degree of dysarthria seen in an individual may vary dependent upon the situation, with stress or excitement exacerbating the dysarthria. Deficits in sentence structures are also sometimes observed.

Traditional Speech Therapy (ie. individual sessions, structured language lessons, articulation therapy) have not been successful in improving communication skills. The use of augmentative communication (manual and electronic) to supplement verbal speech has also had limited success.

The most effective method to enhance communication skills is to provide opportunities for students with LND to successfully communicate in a natural environment. Expanding verbal utterances within a functional setting by allowing the student to become an active participant in daily routines and encouraging self-direction will increase expressive language.

Seating and positioning are also areas to be addressed. Stability and an upright sitting posture will allow for greater phonatory control. With increased phonatory support, there may be some improvement in articulation pattern and intelligibility of speech.

Students with LND exhibit varying degrees of dysphagia (impaired swallowing) and usually require a moist, chopped diet, which may also be necessitated by the extraction of teeth. Oral transport and containment of liquids and solids is generally fair to good. The following suggestions may be useful when feeding:

1. Feed slowly.
2. Alternate liquids and solids.
3. Allow only 2-3 sips of liquid at one time to minimize vomiting.
4. Feed from midline to facilitate swallowing.
5. Keep student in upright position. Tilting backwards increases extension and makes swallowing more difficult.

EDUCATION

There are many challenges facing the Special Education teacher who deals with the Multiply-Handicapped population. These challenges are intensified when the teacher has a student with LND in the class. The educational program for students with LND focuses on a functional curriculum. Student's goals and objectives are directed towards the development of skills in four domains: Community, Vocational, Leisure, and Activities of Daily Living (ADL). A combination of classroom and community based instruction along with the use of adapted equipment provides the experiences necessary for the development of functional skills such as consumer math, communication, self-direction, self-advocacy, and problem solving. It is essential for students to develop an active learning style rather than engage in passive observation.

Due to the nature of the disease, by the time the student is school age, he has most likely acquired "Lesch-Nyhan behaviors" that interfere with learning. Some of the more common behaviors exhibited in school are: screaming, vomiting, cursing, self-injury, oppositional responses (e.g. compulsively giving wrong answers), and manipulating staff through avoidance (saying "I have to go to the bathroom," or "I want a drink of water" or "No more"). These behaviors may occur in a variety of situations such as when there is a pressure to perform (whether in a group, or 1-on-1), during emotional highs and lows due to special events (e.g. field trips, holidays, or home visits), when feeling insecure due to a change in staff or environment, when protective devices are not secure or are inadequate to ensure safety, and when frustrated by the inability to adequately communicate thoughts or feelings. Although behaviors can escalate in these situations, several strategies can be employed to avoid or decrease undesirable behaviors.

The teacher should project confidence in herself or himself and in the student. Students with LND must feel secure in order to function well. If the student senses that the teacher is insecure, and that the situation is poorly controlled and therefore, unsafe, he will become fearful and eventually exhibit "Lesch-Nyhan behaviors."

Communication between the educational staff, family, and support staff must be positive and ongoing. Information regarding injury or illness needs to be communicated promptly so that medical and therapeutic interventions are applied as quickly as possible. Beyond the obvious health reasons for timely interventions, there is the need to demonstrate control of the situation as discussed above, thus avoiding the development of stress and consequent behaviors.

Active participation should be encouraged, for it builds self-confidence and the student is more likely to try new experiences and join in activities. Passive on-looking fosters boredom and increases opportunities for undesirable behaviors.

A major focus of the student's program should be self-direction. The development of self-direction skills builds confidence by giving the student a sense of control over his environment while maximizing independence in all areas.

When a student exhibits a "Lesch-Nyhan behavior," staff should ignore the behavior and redirect or simplify the task. The student should never be allowed to skip his turn and some response should always be elicited. Skipping turns encourages passive on-looking that sets both the teacher and student up for failure by reinforcing avoidance behaviors.

When starting an activity or lesson the teacher should always explain the plan in a step-by-step sequence which lets the student know what is to be expected of him. Once again, the intent is to avoid developing stress.

It is important to start with small tasks that can be performed successfully, and activities should always be completed. If an undesirable behavior is allowed to end an activity, this will reinforce the unwanted behavior and it will most likely occur with greater frequency.

Activities should be conducted in small groups of 2-3 students because students with LND often have difficulty in 1-on-1 activities due to the unavoidable pressure to perform. Too large a group promotes passive on-looking.

At our facility, we have found that traditional academic programs and techniques are generally not very effective with students with LND. Frequently, the pressure to perform in a traditional educational atmosphere elicits "Lesch-Nyhan behaviors" to the point where the student is unable to function as part of a classroom. Instead, approaching academics in functional, real-life settings and activities has been more successful for it encourages active participation, self-direction, and choice-making that a traditional approach does not. Throughout this process, the student is acquiring skills that are necessary to live as independent a life as possible through activities that build self-esteem and lead to tangible successes. For example, a student can work on money skills in the grocery store and succeed by purchasing and carrying away items for use in another area of his program. A good indicator that this approach is beneficial is the number of goals and objectives achieved by students on a regular basis in the functional setting, where these same students were not achieving goals or objectives when the educational format was more traditional.

LEISURE/RECREATION

Recreation therapy and music therapy are beneficial in maximizing an individual's ability to actively participate in a least restrictive environment. Working closely with other disciplines, (e.g. occupational therapy and rehabilitation engineering) individuals with LND are evaluated with regard to interests, physical and cognitive ability, and adapted equipment needs. With individualized adapted equipment a student can participate in a wide variety of activities. When a person with LND participates in recreational or musical activities, various protective devices are employed that provide the necessary security while allowing sufficient range of motion for independent function. For example, when using an electronic keyboard, elbow splints may be used so the student cannot reach his face. The keyboard is well secured to a table at arm's length, and the student can then play without fear of hurting himself or the instrument. Adaptations vary with each student and some individuals require less adaptations than others.

In the leisure setting, as in other environments, students with LND may exhibit subtle forms of self-sabotage. For example, a student playing Nintendo using three switches successfully may eventually decrease to two or one switch, thus decreasing performance and enjoyment. This is interpreted as a form of emotional self-injury and is dealt with accordingly. No criticism is made, nor recognition given to the regression. One tries to find alternative methods of accessing the game to enable continued play. At other times, a student may push a switch off his board or aggressively bang it. Prior to starting the activity it is helpful to have the student direct the placement of the switch. If this behavior still occurs, it is best to ignore

the behavior, pick up the switch and continue with the activity. It may be necessary to find alternate methods of securing the switch (e.g. stronger Velcro, straps, tape, repositioning, etc.). As always, no punishment or other negative feedback is given to the student.

In settings less familiar to the students, such as a new restaurant or grocery store, "Lesch-Nyhan behaviors" become more prevalent due to increased stress. This becomes a learning experience for the individual as well as the community. One tries to facilitate carryover of strategies taught in therapy, and always ensures that some success is achieved. Trips should not be allowed to end prematurely due to a student's behavior. Sometimes it is necessary to take store personnel aside and solicit their cooperation. Community trips can be difficult, but they are very meaningful to the students and should be encouraged.

SUMMARY

Caring for individuals with LND is always a challenge, and without a basic understanding of the unique issues involved in this disease one could easily become overwhelmed. It is hoped that this discussion has provided the basis for at least a beginning knowledge of LND. It must be recognized, however, that even those of us experienced with LND have more questions than answers. There are no state, national, or international organizations that focus on LND to turn to for help, and it is not uncommon for the parents of a boy with LND to report never having met another similar family. When the Matheny School and Hospital recruits new staff, it is assumed that there will be no applicants with prior exposure to children with LND. Working with individuals with LND is a very existential experience. We are essentially on our own. The editors of this Bulletin hope that in addition to providing basic information, we have also provided those readers who are coping as best they can, some degree of comfort in knowing that there are others who have a genuine interest in LND.

REFERENCES

1. Lesch, M., & Nyhan, W. L. A familial disorder of uric acid metabolism and central nervous system function. American Journal of Medicine, 1964, 36, 561-570.

2. Davidson, B., et al. Identification of 17 independent mutations responsible for human hypoxanthine-guanine phosphoribosyl-transferase (HGPRTase) deficiency. American Journal of Human Genetics, 1991, 48, 951-958.

3. Yukaw, T., et al. A female patient with Lesch-Nyhan syndrome. Developmental Medicine and Child Neurology, 1992, 34, 534-546.

4. Crawhall, J. C., et al. Diagnosis and treatment of the Lesch-Nyhan syndrome. Pediatric Research, 1972, 6, 540-543.

5. Silverstein, F. S., et al. Lesch-Nyhan Syndrome: CSF neurotransmitter abnormalities. Neurology, 1985, 35, 907-911.

6. Watts, R. W., et al. Clinical, post-mortem, biochemical, and therapeutic observations on the Lesch-Nyhan Syndrome with particular reference to the neurological manifestations. Quarterly Journal of Medicine, New Series LI, 1982, 201, 43-78.

7. Bakay, B., et al. Experience with detection of heterozygous carriers and prenatal diagnosis of Lesch-Nyhan disease. In M. M. Muller, E. Kaiser, & J. E. Seegmiller (Eds.), Purine Metabolism in Man II: Regulation of Pathways and Enzyme Defects, New York: Plenum Press, 1977, 351-358.

8. Mizuno, T., & Yugari, Y. Prophylactic effect of L-5 hydroxytryptophan on self-mutilation in the Lesch-Nyhan syndrome. Neuropaediatrica, 1975, 6, 13-23.

9. Jolly, D. J., et al. Isolation and characterization of a full length, expressible cDNA for human hypoxanthine guanine phosphoribosyl transferase. Proceedings of the National Academy of Science, 1983, 80, 477.

10. Endres, W., et al. Bone marrow transplantation in Lesch-Nyhan disease. Journal of Inherited Metabolic Disease, 1991, 14, 270-271.

11. Anderson, L., Dancis, J., & Alpert, M. Behavioral contingencies and self-mutilation in Lesch-Nyhan disease. Journal of Consulting and Clinical Psychology, 1978, 46, 529-536.